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Genes and generalists: why we need professionals with added competencies

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INTRODUCTION

Rapid advances of the Human Genome Project are changing medical practice. Over the past two decades, interdisciplinary national panels of physicians, scientists, lawyers, and ethicists have emphasized the central role of genetic counseling in the field of clinical genetics and, thus, a needed clinical skill for physicians to meet the projected demand from patients for genetic tests.

Physicians must, therefore, strengthen their competencies in providing genetic testing to include pretest education and counseling, written informed consent, and posttest counseling of individuals and families. Genetic testing is different from other forms of medical testing or the diagnosis of most adult diseases because, in many cases, the disorder has not yet developed and the results are relevant to other family members.

Unique issues exist in accurately assessing a patient's risk of developing a disease with a genetic predisposition. Some diseases like familial adenomatous polyposis, caused by a mutation of the APC gene, carry a 100% risk for colon cancer and others, such as hereditary breast-ovarian cancer, have a 50% to 80% risk for cancer occurring. Other important aspects of genetic testing include confidentiality of the test information and the psychological effects of the information on individuals and their families. Therefore, becoming skilled in nondirective counseling and recognizing the value of referral to and collaboration with genetics specialists are vital to physicians and their patients.

Summary points

- Rapid advances of the Human Genome Project are changing medical practice
- Genetic services include genetic counseling, genetic testing, and using genetic information to diagnose and treat medical conditions
- Every national consensus group so far has recommended that nondirective genetic counseling be provided with all genetic testing
- National consensus groups have also recommended that some genetic services be provided by specially trained generalist physicians working with genetics specialists because workforce data reveal a projected shortage of genetics professionals
- Generalist physicians need to enhance their skills in obtaining accurate genetic family histories, doing nondirective counseling, and referring patients to and collaborating with genetics professionals

ESSENTIALS OF GENETIC COUNSELING: NATIONAL RECOMMENDATIONS

Sheldon Reed of the Dwight Institute of the University of Minnesota, Minneapolis, in 1947 coined the term "genetic counseling" to describe the type of genetic social work he provided to families: supportive counseling and genetic information about inheritance patterns and recurrence risks without eugenic overtones.[1] In 1975, the Ad Hoc Committee on Genetic Counseling of the American Society of Human Genetics wrote the most widely used definition of genetic counseling (see box).[2]

Genetic counseling can be performed by one person or a team of professionals and should both precede and follow genetic testing. The Institute of Medicine in 1994 made specific recommendations about genetic counseling

Genetic counseling aims to help the individual or family to

- Comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management
- Appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives
- Understand the alternatives for dealing with the risk of recurrence
- Choose the course of action that seems to them appropriate in view of the patient's risk, family goals, and ethical and religious standards and to act in accordance with that decision
- Make the best possible adjustment to the disorder in an affected family member and/or to the risk of that disorder recurring

that reinforced earlier consensus group statements (Table 1).[3] These recommendations reaffirmed that genetic counseling should be nondirective and must be tailored to the cultural perspective of each patient.

A MODEL FOR GENETIC COUNSELING: HEREDITARY BREAST-OVARIAN CANCER

The Human Genome Project is working out the genetic basis of many common diseases of adulthood. The first step toward prevention and treatment is presymptomatic testing and counseling of persons harboring genes that confer susceptibility. Susceptibility testing for hereditary breast-ovarian cancer is an important model for discussing this new era of genetic medicine.

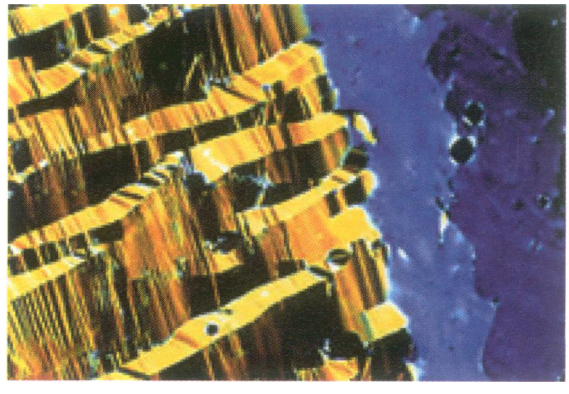
The BRCA1 and BRCA2 genes are tumor-suppressor genes important in regulating the growth of breast epithelial cells; they are responsible for most cases of hereditary breast and ovarian cancer. Few cancers receive the same public attention as breast cancer, in part because it is the second leading cause of death among women aged 25 to 65 years and because the costs are large when the people who develop the disease are young, productive, and often busy raising families and pursuing their careers. The first breast-ovarian cancer susceptibility gene, BRCA1, was isolated in October 1994, and the second such gene, BRCA2, was isolated in December 1995; they are thought to account for about 10% of cases of early-onset, familial breast cancer.[4-6]

In March 1998, the ACP Observer ran a poignant story of a 29-year-old woman with four close relatives, including her aunt and sister, all of whom had had breast cancer or ovarian cancer. The patient approached her newly assigned primary care physician with the desire to receive genetic testing for BRCA1/BRCA2. Her physician, lacking knowledge of the indications for genetic testing, called an oncologist for guidance and obtained information about the commercial laboratory offering genetic testing services. The laboratory required that testing be conducted first with one affected family member, in this case the patient's aunt, followed by testing of the patient and other interested family members. The test results were, in fact, positive for a BRCA1/BRCA2 mutation in the aunt as well as the patient and her sister. The significance of the case was the physician's lack of knowledge and skills in counseling the patient about genetic testing, interpreting the test results, discussing them with the patient and her family members, and collaborating with genetics specialists to develop a management plan. The issue of genetic testing and counseling is, therefore, also important in exposing the need for physicians to acquire new competencies.[7]

Any physician offering BRCA1/BRCA2 genetic testing should be prepared to conduct pretest and post-test counseling that involves a discussion about the natural history of breast cancer, the risk of inheriting a genetic mutation based on the patient's personal cancer history and family history, alternatives to genetic testing—for example, theoretical risk assessment using well-established models[8]—and earlier and more frequent preventive screening with mammography. Physicians should also discuss how the genetic information may be used by the patient in deciding different risk management strategies such as bilateral mastectomy, bilateral oophorectomy, and chemoprevention with tamoxifen citrate, or no intervention. Physicians also must be skilled in obtaining written informed consent, including counseling the patient about possible discrimination in the use of genetic testing information by employers and insurance companies and the psychological effects of the results on the patient and family members.

WHO WILL PROVIDE GENETIC COUNSELING?

Most presymptomatic genetic testing still occurs in research protocols, in which genetic counseling by a qualified genetics professionals is considered mandatory for



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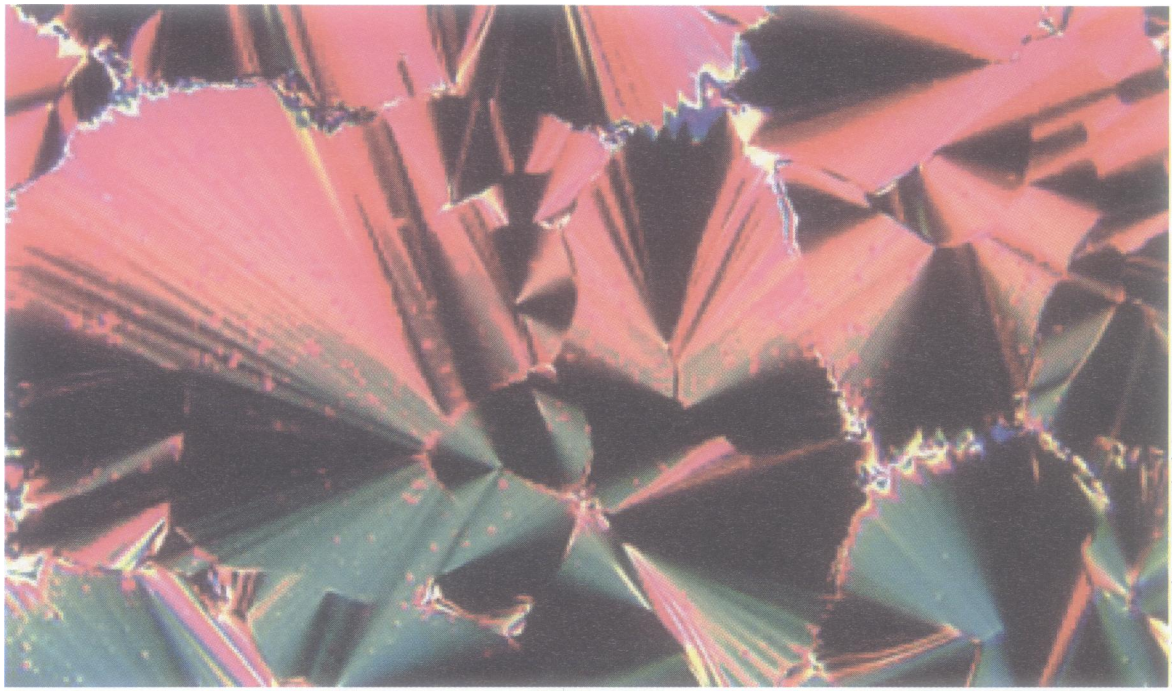
Liquid crystalline DNA

Table 1: *Summary recommendations of national consensus groups*

Principles	The Hastings Center	National Academy of Science	President's Commission	Institute of Medicine
Goals of screening	Improved health in patients with genetic disease Informed reproductive decisions Alleviating anxiety of family at risk	Improved health in patients with genetic disease Informed reproductive decisions Enumeration of genetic disease	Informed reproductive and personal decisions	Informed voluntary reproductive and personal health decisions Screening/testing are viewed as Sources of information
Attainability of goals	Define goals Goals attainable based on pilot studies	Define goals Goals attainable based on pilot studies Standardization of projects	Goals attainable based on pilot studies	Problems should remain investigational until benefits have been defined
Public involvement	Supports community participation in education	Supports public participation Supports involvement of medical community	Supports involvement of medical community	Emphasizes importance of public education Involvement of entire medical community
Access to screening services	Information and screening available for all Priority for high risk groups	Priority for high risk groups	Priority for high risk groups	Equal access and priority to high risk groups, as indicated Recommends expanding third party coverage to the "offering of appropriate genetic testing"
Necessary test characteristics	Precise information to minimize misinterpretation	Acceptable accuracy validity, sensitivity, and specificity	Acceptable accuracy, validity sensitivity and specificity	High test specificity and sensitivity demonstrated through research. Tests to be performed with utmost proficiency and correctly interpreted
Absence of coercion in only obtaining services	Voluntary testing No constraints on childbearing	Voluntary testing	Voluntary testing only newborn screening mandatory only if substantial harm exits	Voluntaries and autonomy are fundamental Mandatory offering of established newborn screening tests
Informed consent	Explicit consent is necessary. Prior to testing clients need to know risks and benefits Ongoing assessment of effectiveness of consent procedure	Explicit consent is necessary Prior to testing clients need to know risks and benefits	Explicity consent is necessary	Full informed consent is an essential element, with delineation of risks, benefits, limitations, other available options, consequences of test results, and disclosure of whether condition tested for is treatable
Protection of subjects	Screening is a form of human experimentation	Screening is a form of human experimentation	Pilot studies are necessary	More research needed, including exploration of psychosocial implications. The only samples that can be used for research purposes are those completely stripped of all identifiers OPRR guidelines to be followed
Disclosure of test results	Full disclosure	Full disclosure	Full disclosure	Full disclosure to individuals tested except in cases of misattributed paternity No disclosure to third parties (insurers/employers) without informed consent
Provision of counseling	Non-directive Define qualifications Ongoing assessment of clients understanding of information and effect information on clients lives	Define qualifications Ongoing assessment of clients understanding of information and effect of information on clients lives	Nondirective Define qualifications Ongoing assessment of clients understanding of information and effect of information on clients lives	Nondirective, patient centered, ethnoculturally sensitive pre-test and post test education and counseling Appropriate trained primary care providers and qualified genetics professionals
Privacy	Information restricted to individual screened	Information restricted to individual screened	Information restricted to individual screened	Confidentiality rigorously protected Information restricted to only individual screened/tested except under rare special circumstances where breach of confidentiality is necessary to avert serious harm
Laboratory provisions	Not specified	Regional facilities Quality control	Regional facilities Quality control	Quality assurance and proficiency program

approval from an institutional review board. As this technology increasingly moves into clinical practice, collaborative teams of genetics professionals, specialists, and primary care physicians will be crucial to offering safe and effective testing. This model of generalists working alongside genetics counselors and geneticists exists throughout the Kaiser Permanente system in California and shows the broad applicability of genetic testing in a managed care population.[9]



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DNA phase transition

Kaiser developed its collaborative model to control costs and to address an increasing shortage of genetics professionals. The American Board of Genetic Counseling recognizes 23 genetic counseling programs at masters level from which about 140 genetic counselors graduate each year.[10,11] At the current level of training, there will be 2,200 genetic counselors at masters level by 2002.[12]

Wilfond and Fost examined the need for professionals trained in genetic counseling in anticipation of large-scale population-based carrier screening for cystic fibrosis.[13] If screening were done annually on 3 million couples who were considering having children, the authors calculated that 561,000 hours would be needed for cystic fibrosis genetic counseling. The Institute of Medicine concluded that generalist physicians must be trained in genetic testing and counseling because the output of genetics professionals will remain inadequate.[3] Hence, initiatives in this rapidly changing field need to be incorporated into continuing medical education.

As genetic tests for conditions with onsets in adulthood continue to emerge in areas such as heart disease and dementia, they will become relevant to a growing number of patients. This will necessitate the development of adjuncts to the labor-intensive and often poorly reimbursed process of pretest education and counseling, such as patient-oriented print material, videotapes, and interactive CD-ROMs. As the public becomes informed, clinicians may be able to spend more time with patients assessing genetic risks, establishing the most effective testing and treatment options, facilitating decision making, and exploring psychosocial issues.

DOCTORS AND PATIENTS: THE CHALLENGING DYNAMICS OF GENETIC COUNSELING

Communication skills and a good physician-patient relationship are important for effective genetic counseling. Emanuel and Emanuel outline four models of the physician-patient relationship. The interpretive model is most appropriate for genetic counseling.[14]

In this interpretive model, the aim of the physician-patient interaction is to elucidate a patient's values and what the patient actually wants. The physician provides information on the nature of the condition and the risks and benefits of possible interventions. Subsequently, the interpretive physicians assist patients in articulating their values and in determining what medical interventions best suit these values. Patients' values are not fixed and may not be known, but often become clear through dialog. This model is also closest to the nondirective model espoused in the genetic counseling literature.[15] In these models, the physician-counselor is more concerned about the process being facilitated than which decision a patient makes.

More recently, Kenen and Smith describe two models of the physician-patient relationship,[16] the first being the mutual participation model advanced in 1956 by Szasz and Hollender.[17] The mutual participation model is most relevant to continuity of care in the primary care setting with the treatment of chronic conditions, where the con-

Table 2

National Society of Genetic Counseling: core competencies for clinicians offering genetic testing

1. Obtain a detailed family history and confirm diagnoses before testing
2. Review the national history of the disease
3. Discuss the predictive value of the test
4. Discuss the risks, benefits, and limitations of testing
5. Explore motivations for requesting testing
6. Discuss readiness for testing and psychosocial issues
7. Describe logistics of testing
8. Present alternatives to testing
9. Discuss confidentiality
10. Outline the process and timing of testing and return of results
11. Provide patient education materials
12. Obtain a written informed consent.

cept of partnership maximizes patients' involvement in decisions and, consequently, enhances compliance to treatment regimens. The goal of the physician is to involve and empower the patient to be an active participant in making decisions about treatment options; patients are encouraged to share concerns so that trust can be established over time.

In the second model, called the "life history narrative model," genetic counselors and client-patients have dual roles. The counselor is both interviewer and listener, and the client is both respondent and narrator. The counselor's main task is to hear the stories that the narrator is telling. The interaction process fosters client power, and the client's beliefs and cognitive perceptions are recognized and valued. With the use of unstructured interviews, clients are encouraged to control the introduction and flow of topics and to openly discuss issues.

This model has been described in the medical encounter literature as patient-centered interviewing, whereby the physician allows the patient to direct the dialog and flow of information.[18,19] The interview begins with the physician asking the patient the reason for the visit and allowing the patient to provide information. Only after the patient has had sufficient time to talk is the physician to enter into the dialog with directed questions about symptoms, illness diagnoses, and treatment options. Research shows that this model of communication enhances empathy and patient satisfaction.[20]

Anticipating the increased use of genetic testing for late-onset disorders in adults, the National Society of Genetic Counselors in 1997 set forth recommendations to assist clinicians in offering genetic testing and counseling.[21] Pretest education and counseling for testing for genetic predisposition for adult-onset conditions, as defined by the National Society of Genetic Counselors, consist of 12 essential steps (Table 2). The National Society of Genetic Counseling further proposed the need for post-test education and counseling to explain the test results and to monitor the need for psychosocial assistance to patients and family members. Generalist physicians now face the challenge of rapidly acquiring these added competencies and integrating them into an already busy clinical practice because the genetic revolution is upon us all.

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